INSTITUTE: National Eye Institute

STUDY NUMBER: 04-EI-0008

PRINCIPAL INVESTIGATOR: J. Fielding Hejtmancik, M.D., Ph.D.

STUDY TITLE: Clinical and Molecular Studies in Families with Corneal Dystrophy or Other Inherited Corneal Diseases

Continuing Review Approved by the IRB on 07/30/15
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Standard

INTRODUCTION

We invite you to take part in a research study at the National Institutes of Health (NIH).

First, we want you to know that:

Taking part in NIH research is entirely voluntary.

You may choose not to take part, or you may withdraw from the study at any time. In either case, you will not lose any benefits to which you are otherwise entitled. However, to receive care at the NIH, you must be taking part in a study or be under evaluation for study participation.

You may receive no benefit from taking part. The research may give us knowledge that may help people in the future.

Second, some people have personal, religious or ethical beliefs that may limit the kinds of medical or research treatments they would want to receive (such as blood transfusions). If you have such beliefs, please discuss them with your NIH doctors or research team before you agree to the study.

Now we will describe this research study. Before you decide to take part, please take as much time as you need to ask any questions and discuss this study with anyone at NIH, or with family, friends or your personal physician or other health professional.

Purpose of the Study

The purpose of this study is to try to identify genes, the basic units of heredity, that are associated with the development of corneal dystrophy or other inherited corneal disease.

Background

You have been invited to participate in this study because you or a member of your family have a condition called a corneal dystrophy or other inherited corneal disease. A corneal dystrophy is a clouding of the cornea in the eye, which passes light to the back of the eye. When the cornea becomes cloudy and doesn't let light through, vision is altered or may even be lost. Corneal dystrophies may occur with vision problems alone, or with other problems, such as changes in facial appearance or bone or joint problems. Corneal dystrophies sometimes run in families, and we believe may be caused by defective genes. Genes, which are made of DNA, tell the cells of your body which proteins to make. When genes are altered, certain cells do not act as they should. In this study, we will try to understand these genetic conditions associated with corneal dystrophies better and to develop better diagnostic tests for corneal dystrophies.

Study Population

Up to 2000 adults and children with corneal dystrophy or other inherited corneal disease and their family members will participate in this study.

Inclusion Criteria

You or your child may be eligible for this study if you or your child have corneal dystrophies or related corneal diseases or if a member of your family has corneal dystrophy or other inherited corneal disease. Participants must be at least 4 years old.

Exclusion Criteria

You may not be eligible for this study if you have corneal dystrophy due to infection, trauma, or another condition that imitates inherited corneal dystrophy. Children may not be eligible if they cannot cooperate with the required eye examinations and blood donation.

Procedures

This study requires 1 visit to the NIH outpatient eye clinic. The visit will last about 3 hours. If all the examinations cannot be done in 1 visit, a 2nd appointment may be needed.

During the study visit, we will review your medical records and ask about your medical and eye disease history and about your family's medical history. The history will be used to draw a family tree. You will have an eye examination and will have a sample of blood drawn for research testing.

The eye examination includes testing how well you see, measuring your eye pressure, and checking your eye movements. For examination of the inside of the eye, your pupil will be dilated with eye drops. While your eyes are dilated we may take pictures of the retina and the inside of your eye. This eye examination is similar to the eye examination you receive from your own eye doctor, but is being done for research purposes only. You will continue to receive eye examinations and treatment from your own doctors while in this study.

To obtain the blood sample, blood will be drawn through a needle in your arm. About 14 cc's (3 teaspoons) of blood will be drawn from adults, less may be drawn from children, depending on their body size.

The blood will be used to try to identify and to study the genes associated with corneal dystrophy in your family. In order to have enough DNA to study, we may create a cell line from your blood cells that will allow us to obtain additional DNA to study in the future without having to draw more blood.

The blood, cell lines, and DNA samples will be stored in secured freezers on the NIH campus or outside laboratories we select for this research project. Samples will be inventoried by codes we assign. The key to the code will be kept in a separate, secure area. These samples will be used for the study described in this consent form.

If a future research project arises where your samples could be useful, we ask you to designate as to whether or not your sample can be used. Any future research use will require approval by the institutional review committees.

Please initial by the line indicating your wishes.

YES, I give permission to use my blood samples in future research studies under the following conditions:

_____These samples may be used for other research projects without contacting me only if the identification code is removed so that the sample can no longer be identified as mine.

_____These samples may be used for other research projects without contacting me even if the code is left on the samples. I understand that if the samples are coded, they may be able to be traced back to my personally identifiable information and my medical records.

_____MAYBE. I wish to be re-contacted if further studies with my samples are considered. After the study has been explained, I will then decide if I want my samples to be included.

_____NO. Under no circumstances shall my samples be used for any future studies. My samples should be discarded once the present study is complete.

If you allow future research on your sample and the research provides information important for your health, we will try to contact you. If you wish to be contacted, please keep the principal investigator for this study, or the NIH Admissions Office, updated about changes in your address or phone number.

If you withdraw from this research project before it is complete, any samples you have contributed will be discarded at the point of your withdrawal. Results obtained prior to your withdrawal from the study will be maintained and your privacy will be protected.

Risks and Discomforts

Risks of Eye Examination, Eye Photography, and Dilation

There is no medical risk from the tests of vision, measuring eye pressure or eye photography. The eye drops used to dilate your eyes may sting. You may have glare and blurry vision for several hours while your eyes are dilated. Some people are allergic to eye drops, while others experience a temporary increase in eye pressure. Your eye could become red or painful. These will be treated if they occur.

Risks of drawing blood

You may have some discomfort and bruising at the site of needle entry. There is a very small risk of fainting or infection in the area of the needle insertion.

Genetic Testing

Genetic testing can provide information about how health or illness is passed on within your family. This knowledge may affect your emotional wellbeing. You might feel differently about your life if you learned that you or your children were at increased risk of a disease, especially if there were no treatment. Your children, brothers or sisters may find out that they are at risk for health problems because of information found out about you, which might affect your relationships with them. Other family members may also be affected by uncovering risks they have but did not want to know about. This information can cause stress, anxiety, or depression.

Some genetic testing can also determine if people are directly related. These tests sometimes show that people were adopted or that their biological parent is someone other than their legal parent. If these facts were not known previously, they could be troubling. Genetic counseling is available at the National Institutes of Health to help you understand the nature and implications of your and your family's genetic findings.

Because of the emotional risk, some people who participate in research do not want to know the results of genetic testing. It is our policy to not disclose the results of genetic testing unless it may have direct medical or reproductive implications for you or your family. You may choose to receive your or your minor child's information or you may choose not to receive the information. Whether you choose to receive the information or not, by agreeing to participate in this study, you do not waive any rights that you may have regarding access to and disclosure of your records. For further information on those rights, you can contact the principal investigator of this study.

Results of genetic testing obtained at NIH are often preliminary and difficult to interpret because the testing is being done for research purposes only and the

laboratories are not clinically certified. Further research may be necessary before these results are meaningful. You may be referred to another laboratory for clinical testing or confirmation. If meaningful information is developed from this study that may be important for your health, you will be informed when it becomes available. If this happens, genetic counseling will be made available to you through the NEI Ophthalmic Genetics Clinic.

Results of genetic testing will become a part of your medical record at NIH. All genetic information is confidential. Medical records containing this information are maintained in a locked and secured manner in the Clinical Center at the National Institutes of Health. Access to your records is limited to authorized NIH staff only. You control the release of any information contained in these files. Genetic information about you will not be revealed to others, including your relatives, without your written permission. Similarly, you will not receive information about other family members. You may only receive information about yourself or your minor children. Parents are entitled to copies of their minor child's medical records without consent of the child.

Problems, such as with insurance or employment discrimination, may occur if you disclose information about yourself or agree to have your research records released. We will not release any information about you or your family to any physician, insurance company or employer unless you sign a document allowing release of the information.

Benefits

You will not benefit directly from participating in this study. We hope, however, to learn more about why some corneal dystrophies affect certain families, which may lead to better diagnosis in the future.

Right of Withdrawal

You are free to withdraw or to refuse study procedures at any time. The investigators may remove you from the study if removal is believed to be in your best medical interest or if you are unable to cooperate with study procedures.

Alternatives to Participation

You will not receive any treatment under this protocol. You may choose not to participate.

Confidentiality

We may publish a chart that shows your family tree and who is affected with the condition, but we will not use your family's name. If you have a unique family, others may still be able to recognize the family tree, but we will use the smallest amount of information possible in order to make the family tree less recognizable.

OTHER PERTINENT INFORMATION

1. Confidentiality. When results of an NIH research study are reported in medical journals or at scientific meetings, the people who take part are not named and identified. In most cases, the NIH will not release any information about your research involvement without your written permission. However, if you sign a release of information form, for example, for an insurance company, the NIH will give the insurance company information from your medical record. This information might affect (either favorably or unfavorably) the willingness of the insurance company to sell you insurance.

The Federal Privacy Act protects the confidentiality of your NIH medical records. However, you should know that the Act allows release of some information from your medical record without your permission, for example, if it is required by the Food and Drug Administration (FDA), members of Congress, law enforcement officials, or authorized hospital accreditation organizations.

2. Policy Regarding Research-Related Injuries. The Clinical Center will provide short-term medical care for any injury resulting from your participation in research here. In general, no long-term medical care or financial compensation for research-related injuries will be provided by the National Institutes of Health, the Clinical Center, or the Federal Government. However, you have the right to pursue legal remedy if you believe that your injury justifies such action.

3. Payments. The amount of payment to research volunteers is guided by the National Institutes of Health policies. In general, patients are not paid for taking part in research studies at the National Institutes of Health. Reimbursement of travel and subsistence will be offered consistent with NIH guidelines.

4. Problems or Questions. If you have any problems or questions about this study, or about your rights as a research participant, or about any research-related injury, contact the Principal Investigator, J. Fielding Hejtmancik, M.D. (301) 496-8300; TDDY (301) 402-4175. Nights & Weekends: (301) 496-5748.

You may also call the Clinical Center Patient Representative at (301) 496-2626.

5. Consent Document. Please keep a copy of this document in case you want to read it again.

COMPLETE APPROPRIATE ITEM(S) BELOW:

A. Adult Patient's Consent

I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby consent to take part in this study.

Signature of Adult Patient/Legal Representative _____ Date _____

Print Name _____

B. Parent's Permission for Minor Patient.

I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby give permission for my child to take part in this study. (Attach NIH 2514-2, Minor's Assent, if applicable.)

Signature of Parent(s)/Guardian _____ Date _____

Print Name _____

C. Child's Verbal Assent (If Applicable)

The information in the above consent was described to my child and my child agrees to participate in the study.

Signature of Parent(s)/Guardian _____ Date _____ Print Name _____

THIS CONSENT DOCUMENT HAS BEEN APPROVED FOR USE FROM JULY 30, 2015 THROUGH JULY 29, 2016.

Signature of Investigator _____ Date _____

Print Name _____

Signature of Witness _____ Date _____

Print Name _____